

WHAT IS CLAIMED IS:

1. A method of characterizing a chromosomal abnormality in a fetus by performing a comprehensive biochemical analysis of a specimen of amniotic fluid comprising:

5 obtaining a comprehensive profile of metabolites in the specimen of amniotic fluid,
comparing the profile with a control profile of metabolites that is representative of normal levels of metabolites,

analyzing the profile with respect to the normal profile by identifying each metabolite that has a different level when compared with the normal level of that metabolite,

generating a biochemical characterization of the abnormality, and

prescribing a biochemical treatment for each metabolite that has a different level when compared with the normal level of that metabolite.

2. The method of Claim 1 wherein comparing the profile with respect to the normal profile is accomplished by comparing mean levels and standard deviations for each metabolite.

3. The method of Claim 1 wherein comparing the profile with respect to the normal profile is accomplished by comparing median levels using a nonparametric analysis for each metabolite.

4. The method of Claim 1 wherein Down Syndrome is the chromosomal abnormality that is diagnosed.

5. The method of Claim 1 wherein the metabolite is chosen from the group consisting of organic acids, amino acids, neurotransmitters, fatty acids, glycine conjugates, drugs, drug metabolites, hormones, vitamins, and carbohydrates.

5 6. The method of Claim 1 wherein the metabolites comprise multiple categories of metabolite groups that are analyzed simultaneously.

7. A method of performing a comprehensive biochemical analysis of a specimen of amniotic fluid in order to characterize a chromosomal abnormality in a fetus comprising:
obtaining a comprehensive profile of metabolites in the specimen of amniotic fluid,
comparing the profile with a control profile of metabolites that is representative of normal levels of the reported metabolites,
analyzing the profile with respect to the normal profile by identifying each metabolite that has a different level when compared with the normal level of that metabolite,
inferring an activity level for an enzyme that corresponds to the identified metabolite,
inferring a cofactor level based on the activity level for the enzyme,
generating a global biochemical characterization of the abnormality, and
prescribing a biochemical treatment for each metabolite that has a different level when compared with the normal levels.

8. A method of characterizing a chromosomal abnormality in a fetus by performing a comprehensive biochemical analysis of a specimen of amniotic fluid comprising:
obtaining a comprehensive profile of metabolites in the specimen of amniotic fluid,

comparing the profile with a control profile of metabolites that is representative of levels of metabolites in patients suffering from the chromosomal abnormality,

analyzing the profile with respect to the chromosomal abnormality profile by identifying each metabolite that has a same level when compared with the abnormal level of that metabolite,

5 and

prescribing a biochemical treatment for each metabolite that has a same level when compared with the abnormal level of that metabolite.

9. The method of Claim 8 wherein comparing the profile with respect to the abnormal profile is accomplished by comparing mean levels and standard deviations for each metabolite.

10. The method of Claim 8 wherein comparing the profile with respect to the abnormal profile is accomplished by comparing median levels using a nonparametric analysis for each metabolite.

11. The method of Claim 8 further comprising:
inferring an activity level for an enzyme that corresponds to an identified metabolite having a same level as a metabolite in the abnormal profile, and

20 inferring a cofactor level based on the activity level for the enzyme.

add C12
add B2

Add C1